



Exon-Intron Boundary Sequences of the Human alpha-7 nAChR Subunit Gene

Exon Number	Exon length (bp)	cDNA position	splice acceptor	flanking exon sequence	splice donor	Intron number	Intron Size approximate (Kb)
1	55	1-55		...CTG CAC G L H	GTAAGGCCAC	1	0.3
2	140	56-195	TCTCCTTAAG	TG TCC.....GAC GTG V S D V	GTGAGTCCCG	2	Unknown
3	45	196-240	TTTTTTTGAAG	GAT GAG.....CAA ATG D E Q M	GTAAGTTAAG	3	9.0
4	110	241-350	TGTGTGTCAG	TCT TGG.....AAC AG S W N S	GTAAGCATAT	4	Unknown
5	80	351-430	CTGTTTCTAG	T GCT GAT.....CCT CCA G A D P P	GTAAGCTGCA	5	4.0
6	168	431-598	ACCCACACAG	GC ATA.....CTA GTG G G I L V	GTAAGCCATG	6	1.0
7	195	599-793	CCCTATGGAG	GA ATC.....TCC CTG G G I S L	GTAAGCGCCC	7	1.0
8	87	794-880	TATGTTTTAG	GG ATA.....TTG ATA G G I L I	GTAAGGCAAG	8	3.5
9	110	881-990	CTCTCCACAG	CC CAG.....AAG TGG A Q K W	GTACGTTCTT	9	5.0
10	519	991-1509	GTCTCCCCAG	ACC AGA... T R			

FIG. 1



Sequence Variants Identified in Full-Length and Duplicated Genomic Clones

DNA	EXONS CONT.	EXON 6 +/- 497-498	EXON 7 654	EXON 7 690	EXON 10 1269	EXON 10 1335	L76630
CHR15 HYBRID	5-10 1-10	+TG -TG	C/T	G/A	C/C	C/C	6GT 8GT
YAC							
D-948a10	5-10	-TG	T	A	C	C	6GT
D-853b12	6-10	-TG	T	A	C	C	6GT
D/F 969b11	5-10 1-10	+TG -TG	C/T	G/A	C/T	C/C	6GT 8GT
F-134h10	1-10	+TG	C	G	C	C	8GT
F-776a12	1-10	+TG	C	G	C	C	8GT
F-791e6	1-10	+TG	C	G	C	C	8GT
F-811b6	1-10	+TG	C	G	C	C	8GT
F-953g6	1-10	+TG	C	G	C	C	8GT
F-859c11	1-10	+TG	C	G	C	C	8GT
F-810f11	1-10	+TG	C	G	C	C	8GT
F-801e1	1-10	+TG	C	G	C	C	8GT
BAC							
F-467o18	1-10	+TG	C	G	C	T	8GT

FIG. 2

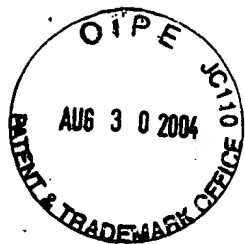
DNA	Control #	EXON 6 +/- 497-498	EXON 7 654	EXON 7 690	EXON 10 1269	EXON 10 1335
Control Genomic DNA	43	+/+ 10 +/- 33 -/- 0	C/C 5 C/T 38 T/T 0	G/G 0 G/A 43 A/A 0	C/C 6 C/T 36 T/T 1	C/C 24 C/T 19 T/T 0



# Expression Analysis of Sequence Variants

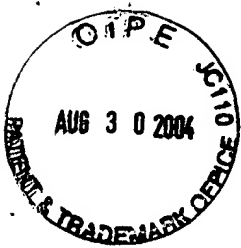
Subj	Bases 497-498			Base 654			Base 690			Base 933			Base 1296			Base 1335		
	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA
SL061	+/-	+	+/-	CT	C	CT	GA	G	GA	G	G	G	CT	CT	CT	C	C	C
SL084	+	+	+	C	C	C	GA	G	GA	G	G	G	CT	C	CT	C	C	C
SL111	+/-	+	+/-	CT	C	CT	GA	G	GA	G	G	G	CT	CT	CT	CT	CT	CT
SL097	+	+	+	CT	C	CT	GA	G	GA	G	G	G	CT	C	CT	C	C	C
SL089	+	+	+	C	C	C	GA	GA	GA	GA	GA	GA	CT	CT	CT	C	C	C
SHSY	+/-	+	+/-	CT	C	CT	GA	GA	GA	GA	GA	GA	C	C	C	C	C	C

FIG. 3



-392 agaacgcaag ggagaggtag agcctggcct tgggcagccc ctggcctggc cagagggcgc aggccgagag  
AP-2  
-322 cccgctcggt ggagactggg ggtggagggtg cccggagcgt acccagcgc gggagtagct cccgctcaca  
-252 cctcgggctg cagttccctg ggtggccgc gagacgctgg cccgggctgg agggatggcg gggcggggac  
-182 gggggcgggg gcggggctcg tca<sup>CREB</sup>gtggag aggcgcgcgg gggc<sup>Sp1</sup>gggcgg cgcgcgccgc  
-112 tccttaaagg cgcgcgagcc ggcgcgcgag gtgcctctgt ggcgcgcagg gcaggcccg ggcacagccg  
-42 agacgtggag cgcgcgggt cgtgcagct ccgggactca acATGCGCTG CTCGCCGGA GCGCTCTGGC  
Met  
+29 TGGCGCTGGC CCGCTCGCTC CTCACGgta aagccac

FIG. 4



CENTROMERE

TELOMERE

PAC BAC YAC	SIZE kb	ALPHA-7 SEQUENCE										ALPHA-7 SEQUENCE									
		D1581043	D158942	D1581043	L76630	EXON 9	EXON 8	EXON 7	EXON 6	EXON 5	EXON 4	EXON 3	EXON 2	EXON 1	D1581360	D1581010	D158144	D1581007	D158995		
PAC																					
64a1		-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
25919		-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
BAC		-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
467o18		-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
YAC																					
948a10	1730	+	+	+	+	+	+	+	+	+	+	+	+	+	-	-	-	-	-	-	-
853b12	790	+	+	+	+	+	+	+	+	+	+	+	+	+	-	-	-	-	-	-	-
895f6	1580	+	+	+	+	+	+	+	+	+	+	+	+	+	-	-	-	-	-	-	-
969b11	1030	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
776a12	1640	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
791e6	1170	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
811b6	1060	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
953g6	1720	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
134h10	N.A.	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
859c11	1330	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
810f11	940	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
801e1	1630	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
966a4	1500	-	-	-	-	-	-	-	-	-	-	-	-	-	+	-	-	-	-	-	-
764f8	740	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-
822g2	1280	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-

FIG. 5



<b>EXON D</b> 297bp	1 CAGGCCGCCA CAGCGCGCG AGCGGCTCGG TTCAAGGCCA CCCACCGCAA	CATAGCTCCC GGGAGAGGTG GACTGGCTCC AACCGAAGTT CATTAAGgtg	GCCAAGTCCT GAGCCGCGAG TTTTCCGCGC ACTGGCCTCT agtgcgc.....	CGGTGCCCCCT AGCTCGGCCG CCCTCCCCGCC ATCTTCCAGG agtgcgc.....	TGCCATTTTTC GGGGCCCCCG GGAGGTGAGG AGAACCAGGA agtgcgc.....	CAGCCGCGTC CTGGTGGCCG GGAAGATGTC GCCACAGCCG agtgcgc.....	CCACGAGGGT CGGCCATGAC CATGTCAGGG CGGCTCACGC agtgcgc.....
<b>EXON C</b> 125bp	.....ctc TATCTACACG AAgtgagttg 422	atttcagATT ACTCAGATCT ta.....	ACAAGTGGAC TGTGTGCACC ta.....	ACCTGAGTCA CCCATTATTG ta.....	GCAGGACCTG ACAATCCAAA ta.....	GAATCCCAGA GGTGCAGAAA ta.....	TGAGAGAGCT GCACTCTGAC ta.....
<b>EXON B</b> 64bp	...ttaaccac TTTCAGgtag 486	agATAATGAA gateat.....	ACAACCACCA ta.....	TCGGTTAAAT ta.....	TTGATGCAAA ta.....	AATATTGCAT ta.....	CTACCAGCAT ta.....
<b>EXON A</b> 47bp	.....ttta aca.....	ttctagTTCC ta.....	AATTGCTAAT ta.....	CCAGCATTTC ta.....	TGGATAGCTG ta.....	CAAACGCGA 533	TATgtaagta 533
<b>EXON 5</b> 80bp	...ctgtttc ta.....	tagTGCTGAT GCATTGCCAG	GAGCGCTTTG TACCTGCCTC	ACGCCACATT CAGgtaagctgca.....	CCACACTAAC 613	GTGTGTGGTGA 613	ATTCTTCTGG 613
<b>EXON 6</b> 27bp	....accacaca ta.....	cagGCATATT ta.....	CAAGAGTTCC ta.....	TGCTACATCG 640	ta.....	ta.....	ta.....

FIG. 6

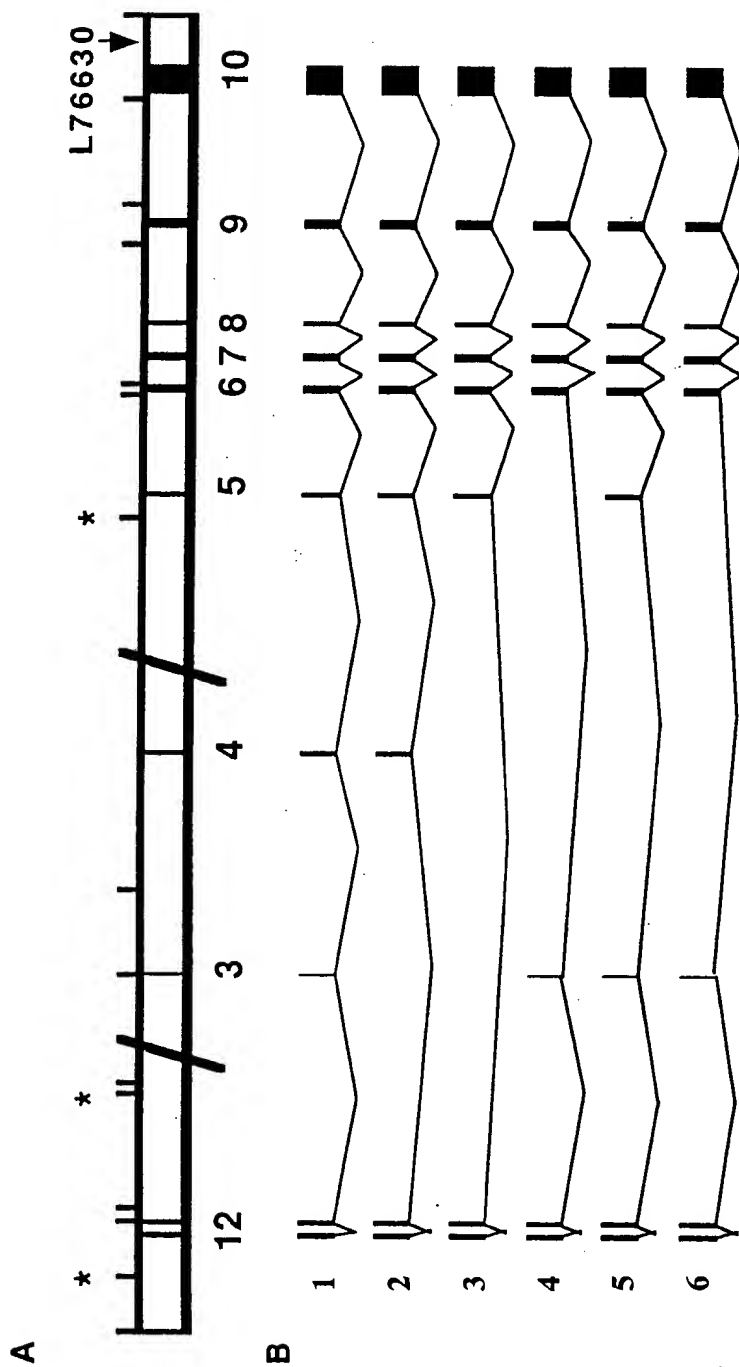


FIG. 7



1 agaacgcaag ggagaggtag agcctggcct tgggcagccc ctggcctggc cagaggcgcg  
61 aggccgagag cccgctcggt ggagactggg ggtggaggtg cccggagcgt acccagcgcc  
121 gggagtacct cccgctcaca cctcgggctg cagttccctg ggtggccgcc gagacgctgg  
181 cccgggctgg agggatggcg gggcggggac gggggcgggg gcggggctcg tcacgtggag  
241 aggcgcgcgg gggcggggcg ggcggggggc cgcgcccggc tccttaaagg cgcgcgagcc  
301 gagcggcgag gtgcctctgt ggccgcaggc gcaggcccgg gcgacagccg agacgtggag  
361 cgcgccggct cgctgcagct ccgggactca ac

FIG. 8

1 caggccgcca catagctccc gccaaagtcc cggtgcccct tgccattttc cagccgcgct  
61 cccacgaggg tcacggcgcc ggggagaggt ggagccgcga gagctcggcc gggggccccc  
121 cctggtggcc gcggccatga cagcggctcg ggactggctc cttttccgcg cccctcccgc  
181 cggaggtgag gggaagatgt ccatgtcagg gttcaaggcc aaaccgaagt tactggcctc  
241 tatcttccag gagaaccagg agccacagcc gcggctcacg ccccaccgca acattaagat  
301 tacaagtgga cacctgagtc agcaggacct ggaatcccag atgagagagc ttatctacac  
361 gactcagatc ttgttgtcac ccccattatt gacaatccaa aggtgcagaa agcactctga  
421 caaataatga aacaaccacc atcgggttaa tttgatgcaa aaatattgca tctaccagca  
481 ttttcagttc caattgctaa tccagcattt gtggatagct gcaaactgcg atattgctga  
541 tgagcgcttt gacgccacat tccacactaa cgtgttggtg aattcttctg ggcattgcca  
601 gtacctgcct ccaggcatat tcaagagttc ctgctacatc g

FIG. 9

1 agccctttcc caggcggtag cgggggcagt ggtgctgttg cccttttaaa ctgcggcttg  
61 acgggagccg cgcctcctgt cgggtggagtc ggttataaaag ggagcagccc cgcaggccgc  
121 cacatagctc ccgccaagtc ctcggtgccc cttgccattt tccagccgcg ctcccacgag  
181 ggtcacggcg gcggggagag gtggagccgc gagagctcgg ccggggggccc cgcctggtgg  
241 ccgcggccat gacagcggct cgggactggc tccttttccg cgccccctcc gccggaggtg  
301 aggggaagat gtccatgtca gggttcaagg ccaaaccgaa gttactggcc tctatcttcc  
361 aggagaacca ggagccacag ccgcggctca cgccccaccg caacattaag attacaagtg  
421 gacacctgag tcagcaggac ctggaatccc agatgagaga gcttatctac acgactcaga  
481 tcttgttgtc acccccatta ttgacaatcc aaaggtgcag aaagcactct gacaattcca  
541 attgctaadc cagcatttgt ggatagctgc aaactgcgat attgctgatg agcgctttga  
601 cgccacattc cacactaacg tgttggtgaa ttcttctggg cattgccagt acctgcctcc  
661 aggcataattc aagagttcct gctacatcg

FIG. 10